

**Disease**

# Carbonic anhydrase VA deficiency<sup>1, 2</sup>

**Gene**

## CA5A

|                                                 |                                                                                                                                                                                                                         |
|-------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                                                      |
| <b>Current Treatments</b>                       | sodium benzoate, N-carbamylglutamate, limit protein with illness                                                                                                                                                        |
| <b>Evidence for Treatment</b>                   | expert opinion                                                                                                                                                                                                          |
| <b>Non-Molecular Testing Availability</b>       | no                                                                                                                                                                                                                      |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=ca5a&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=ca5a&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

**References**

<sup>1</sup> <https://www.ncbi.nlm.nih.gov/books/NBK284774/#ca5a-def.Summary>

<sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/32809955/>